Translocation t(11;22) in Esthesioneuroblastoma

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ABSTRACT: *Esthesioneuroblastoma is an exceedingly rare malignant neuroectodermal tumor of olfactory epithelium origin. We have performed cytogenetic studies on a tissue culture line established from a metastatic lesion in one such patient and observed that, among several chromosomal abnormalities, the cells contained a reciprocal translocation, t(11;22)(q24;q12), indistinguishable from the one that has been reported in Ewing's sarcoma, Askin's tumor, and peripheral neuroepithelioma. The uniqueness of this marker suggests that these tumors may be derived from the same type of stem cell, with varying histopathologic and clinical manifestations.*

INTRODUCTION

Esthesioneuroblastoma is a malignancy that originates in the neuroectodermal stem cells of the olfactory epithelium; it is exceedingly rare [1–3]. Although initial local therapy often results in a permanent cure, local recurrence with intracranial extension or distant metastases are common and eventually result in death [4]. We have performed cytogenetic studies on a tissue culture line established from a metastatic lesion in one such patient and observed that, among several chromosomal abnormalities, the cells contained a reciprocal translocation, t(11;22)(q24;q12), indistinguishable from the one that has been reported in Ewing's sarcoma, Askin's tumor, and peripheral neuroepithelioma [5].

CASE REPORT

The patient was a 22-year-old Caucasian male who presented with epitaxis, nasal stuffiness, and headaches in October 1982. On examination, a large polyploid lesion was found to extend from the superior nasal cavities into both maxillary sinuses. He underwent turbinectomy, septectomy, and bilateral Caldwell–Luc resection; all gross disease was removed from the maxillary sinuses and 5600 rad in 28 fractions were delivered locally. The patient was in complete remission until a metastatic lesion, clinically presenting as a swelling of the left anterior chest, was observed. Computed tomographic scan of the thorax revealed a 10 × 10 cm mass in the chest

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wall with invasion of the fifth and sixth ribs. He was then referred to the NCI/Navy Medical Oncology Branch and biopsy of a mass on the right lower chest wall on March 20, 1985 confirmed the diagnosis of recurrent esthesioneuroblastoma. Neural origin of the tumor was demonstrated by the presence of dense core neurosecretory granules in electromicroscopy preparations. A cell line was established, and within 1 year cytogenetic studies of 30 adherent, nonfloating cells revealed a hyperdiploid clone with a 47,XY,+12,−18,+del(8)(q12),t(8;17)(q12;p12),t(11;22)(q24;q12) karyotype in 70% of the cells (Fig. 1); the remaining 30% were also hyperdiploid but contained a t(2;14)(p25;p12) marker instead of the del(8)(q12) and t(8;17) markers. All cells contained the t(11;22) marker.

DISCUSSION

Esthesioneuroblastoma is an uncommon malignancy and no cytogenetic studies have been reported heretofore. In a recent publication, we reported the observation of a common chromosomal marker, t(11;22)(q24;q12), in Ewing's sarcoma, Askin's tumor, and peripheral neuroepithelioma, all of which show evidence of neural origin. The uniqueness of the marker suggests that these tumors may be derived from the same type of stem cell, with varying histopathologic and clinical manifestations.

Wade et al. [4] studied the response to chemotherapy in five patients with ad-

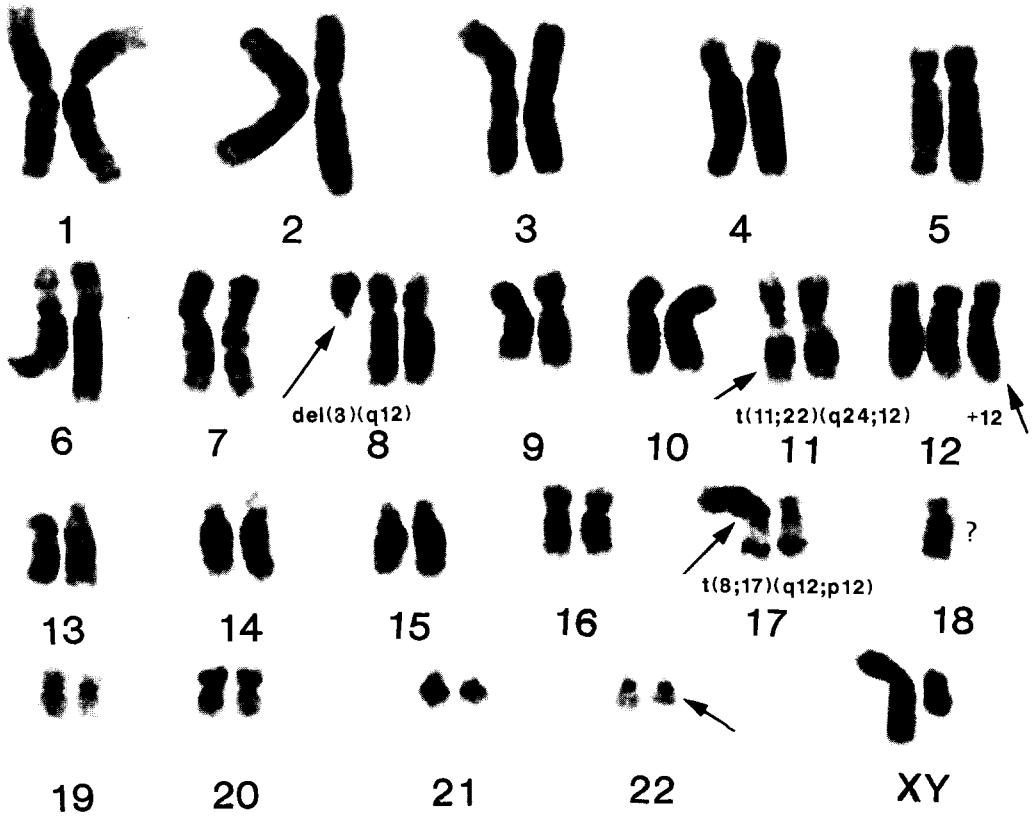

Figure 1 G-banded karyotype of a metaphase from an esthesioneuroblastoma cell line showing the t(11;22)(q24;q12) marker and other chromosomal abnormalities 47,XY,+12,−18,+del(8)(q12),t(8;17)(q12;p12),t(11;22)(q24;q12).

vanced stage or metastatic esthesioneuroblastoma and reviewed an additional eight patients from the literature; eight of the 13 patients (62%) had an objective response to chemotherapy. Because chemotherapy is effective in treating Ewing's sarcoma [6], a tumor with an identical chromosome translocation, it is suggested that the same chemotherapeutic agents may be useful in esthesioneuroblastoma, as well.

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